

Report of the committee on nomenclature

Prepared by:  
E. R. Giblett

Committee members:

E. R. Giblett, Seattle (chairman)  
H. Harris, London  
P. Meera Khan, Leiden  
E. W. Lovrien, Portland

W. J. Mellman, Philadelphia  
C. W. H. Partridge, New Haven  
T. B. Shows, Buffalo

The guidelines outlined below are based on the decisions made at an interim meeting of the committee held on April 3-4, 1975. These guidelines, including a list of suggested names for enzyme loci, were approved during a plenary session of the Gene Mapping Workshop in Baltimore, 1975.

Consideration was confined to enzyme nomenclature because the blood group antigens have been dealt with in the standard text by Race and Sanger (1), while participants in the Histocompatibility Testing workshops have handled the terminology problems in that area (2). In general, the guidelines for naming enzymes can also be applied to the plasma proteins.

Guidelines for Genetic Nomenclature of Human Enzymes

Essential to a satisfactory terminology are that it be precise and unambiguous, /it clearly distinguish between genotypes and phenotypes, and/as far as possible, the symbols used should readily identify the particular enzyme. In addition it should be sufficiently flexible to permit some unusual symbols/used to designate certain enzymes in the original papers and have subsequently been widely adopted in the literature. It should also be capable of incorporating new discoveries as they are reported.

The following general scheme appears to meet these requirements and is reasonably convenient in practice.

### I. Genotypes

Genotypic symbols, i.e. for loci or alleles, are italicised (or underlined in typescript) to distinguish them clearly from symbols used to designate phenotypes, which are not italicised or underlined.

#### A. Loci

1. Loci are designated by letters, either all capitalized (preferred) or just the first letter. Usually two or three letters will suffice, but sometimes four or even five may be required.

Examples: ADA for adenosine deaminase

UMPK for uridine monophosphate kinase

Gd for glucose-6-phosphate dehydrogenase

(Obviously, when lower case letters are used to designate one locus, it is undesirable to use the same letters but in capitals (e.g. GD) to designate another locus.) The letters chosen for locus names are preferably based on the recommended name given by the Enzyme Commission on Nomenclature. However, this is sometimes inconvenient or confusing because of past usage. Thus, GOT is preferred for glutamic-oxaloacetic transaminase, although the E.C. recommended name is aspartate aminotransferase. In some cases, Greek letters are also needed for clarity. Example:  $\alpha$ GAL for  $\alpha$ -galactosidase to distinguish it from  $\beta$ -galactosidase ( $\beta$ GAL).

2. There are often two or more loci coding for different polypeptide chains which are contained in separate enzyme proteins having very similar or identical catalytic properties. Such loci are best differentiated by appropriate subscripts.

Examples:

PGM<sub>1</sub>, PGM<sub>2</sub> and PGM<sub>3</sub> for the three phosphoglucomutase loci

ADH<sub>1</sub>, ADH<sub>2</sub> and ADH<sub>3</sub> for the three alcohol dehydrogenase loci

Although numerical subscripts are often most convenient, sometimes because of past usage or ease/identification, letters are preferred to avoid confusion.

Examples:

LDH<sub>A</sub>, LDH<sub>B</sub> and LDH<sub>C</sub> for the three lactate dehydrogenase loci

PGAM<sub>M</sub> and PGAM<sub>B</sub> for the two phosphoglycerate mutase loci which are active in muscle and brain, respectively.

Some enzymes occur in a so-called soluble (or supernatant or cytosol) form and also in a mitochondrial form, with the two forms being catalytically similar but coded at separate loci.

In such cases, the use of S and M as subscripts may be less confusing than numerical or alphabetical designations.

Example:

GOT<sub>S</sub> and GOT<sub>M</sub> for the soluble and mitochondrial forms of glutamic-oxaloacetic transaminase.

B. Alleles

Different alleles at the same locus are designated by superscripts.

Example:

PGM<sup>1</sup>, PGM<sup>2</sup>, PGM<sup>3</sup>, PGM<sup>4</sup> etc., for alleles at the PGM<sub>1</sub> locus.

The superscripts may be numerical or alphabetical. In rare cases, + and - signs, when used extensively in the past, may be retained.

Example:

Gd<sup>B</sup>, Gd<sup>A</sup>, Gd<sup>A-</sup> for the three common alleles at the glucose-6-phosphate dehydrogenase locus in Black populations.

In other cases, place names are best used as the allele superscript to avoid confusion.

Example:

Gd<sup>Mediterranean</sup>, Gd<sup>Canton</sup>, Gd<sup>Athens</sup>, Gd<sup>Seattle</sup>

(Abbreviation of the place name may be more convenient.)

So-called "null" or "silent" alleles with little or no associated enzyme activity are best designated by the superscript 0 (i.e. zero), although the letter s may be retained because of common usage.

Examples:

PGM<sup>0</sup>, E<sup>s</sup><sub>1</sub> ("silent" allele of the serum cholinesterase first locus)

When heterogeneity between "null" alleles can be demonstrated, the allele designation should be qualified, as by a place name.

Example:

ADA<sup>0</sup> Calcutta

C. Examples of Genotypes

The following are some typical examples of genotypes written in accordance with the above recommendations and section D (below).

1. Heterozygote for the two common alleles at the ADA locus:

ADA<sup>1</sup>ADA<sup>2</sup> (or ADA<sup>1</sup>/ADA<sup>2</sup>)

2. Heterozygotes for one or the other of these common ADA alleles and a "null" allele not separable from other "null" alleles at this locus:

ADA<sup>1</sup>ADA<sup>0</sup> and ADA<sup>2</sup>ADA<sup>0</sup> (or ADA<sup>1</sup>/ADA<sup>0</sup> and ADA<sup>2</sup>/ADA<sup>0</sup>) .

3. Genotype of an individual heterozygous for the two common alleles of PGM<sub>1</sub>, homozygous for the common allele of PGM<sub>2</sub> and heterozygous for the two common alleles of PGM<sub>3</sub> (3 unlinked loci):

PGM<sub>1</sub><sup>1</sup>/PGM<sub>1</sub><sup>2</sup>, PGM<sub>2</sub><sup>1</sup>/PGM<sub>2</sub><sup>1</sup>, PGM<sub>3</sub><sup>1</sup>/PGM<sub>3</sub><sup>2</sup>

or

|                                      |                                      |                                      |
|--------------------------------------|--------------------------------------|--------------------------------------|
| <u>PGM</u> <sub>1</sub> <sup>1</sup> | <u>PGM</u> <sub>2</sub> <sup>1</sup> | <u>PGM</u> <sub>3</sub> <sup>1</sup> |
| <u>PGM</u> <sub>1</sub> <sup>2</sup> | <u>PGM</u> <sub>2</sub> <sup>1</sup> | <u>PGM</u> <sub>3</sub> <sup>2</sup> |

D. Linkage and Phase

A slash, either horizontal or semivertical (— or /) separating alleles, implies chromosomal location. The slash may be omitted in designating the genotype at a single locus. However, if two or more loci are involved, a horizontal line is recommended, particularly if the loci are syntenic.

1. Non-syntenic loci may be designated either by an interrupted horizontal line or by individual slashes and separation by commas.

Example:

$\frac{\text{ADA}^1}{\text{ADA}^2}$      $\frac{\text{PGM}^1}{\text{PGM}^2}$     or     $\text{ADA}^1/\text{ADA}^2$ ,     $\text{PGM}^1/\text{PGM}^2$

2. When the loci are in the same linkage group and the phase is known, the horizontal line is continuous.

Example:

$\frac{\text{AMY}_1^A}{\text{AMY}_1^B}$      $\frac{\text{AMY}_2^B}{\text{AMY}_2^A}$     (i.e.  $\text{AMY}_1^A$  and  $\text{AMY}_2^B$  are in *cis* position, as are their alleles)

3. When the loci are in the same linkage group but the phase is not known, a semicolon is used.

Example:

$\frac{\text{AMY}_1^A}{\text{AMY}_1^B}$  ;  $\frac{\text{AMY}_2^A}{\text{AMY}_2^B}$

4. To designate loci which are syntenic but not in the same linkage group, a colon is used.

Example:

$\frac{\text{AMY}_2^A}{\text{AMY}_2^B} : \frac{\text{PGM}_1^1}{\text{PGM}_1^2}$

## II. Phenotypes

- A. The phenotypic designation should have the same letters and subscripts as the locus (but not italicised or underlined), followed by the numerical, alphabetical or other symbol for the alleles, but not as superscripts. In the case of homozygotes for any allele or heterozygotes for a "null" allele, only one allele symbol is used.

Examples:

| <u>Genotype</u>   | <u>Phenotype</u>  |
|---|---|
| <u>ADA</u> <sup>1</sup> <u>ADA</u> <sup>1</sup>   | ADA 1   |
| <u>ADA</u> <sup>1</sup> <u>ADA</u> <sup>2</sup>   | ADA 2-1   |
| <u>ADA</u> <sup>2</sup> <u>ADA</u> <sup>2</sup>   | ADA 2   |
| <u>ADA</u> <sup>1</sup> <u>ADA</u> <sup>0</sup>   | ADA 1   |
| <u>ADA</u> <sup>2</sup> <u>ADA</u> <sup>0</sup>   | ADA 2   |
| <u>PGM</u> <sub>1</sub> <sup>1</sup> / <u>PGM</u> <sub>1</sub> <sup>2</sup> , <u>PGM</u> <sub>2</sub> <sup>1</sup> / <u>PGM</u> <sub>2</sub> <sup>1</sup> , <u>PGM</u> <sub>3</sub> <sup>1</sup> / <u>PGM</u> <sub>3</sub> <sup>2</sup> | <u>PGM</u> <sub>1</sub> 2-1, <u>PGM</u> <sub>2</sub> 1, <u>PGM</u> <sub>3</sub> 2-1 |

For hemizygotes, heterozygotes and homozygotes of the X-linked phosphoglycerate kinase alleles PGK<sup>1</sup> and PGK<sup>2</sup>,

| <u>Genotype</u>                                 | <u>Phenotype</u> |
|---|------------------|
| <u>PGK</u> <sup>1</sup>                         | PGK 1            |
| <u>PGK</u> <sup>2</sup>                         | PGK 2            |
| <u>PGK</u> <sup>1</sup> <u>PGK</u> <sup>1</sup> | PGK 1            |
| <u>PGK</u> <sup>1</sup> <u>PGK</u> <sup>2</sup> | PGK 2-1          |
| <u>PGK</u> <sup>2</sup> <u>PGK</u> <sup>2</sup> | PGK 2            |

**III. Isozyme Subunits**

When two or more loci code for different polypeptide chains which occur together as subunits of single isozymes in a set of isozymes, it is useful to designate the subunit structure of the individual isozymes.

Greek letters are convenient symbols for the polypeptide chains. A different letter can be used for the peptide product of each locus, by analogy with the  $\alpha$ ,  $\beta$ ,  $\gamma$  and  $\delta$  chains of hemoglobin. Whenever there are two or more alleles at a given locus coding for structurally different forms

of the same polypeptide, superscripts are incorporated which are the same as the superscripts used to designate the corresponding alleles.

Example:

The three loci of alcohol dehydrogenase, ADH<sub>1</sub>, ADH<sub>2</sub> and ADH<sub>3</sub> are thought to code for three different polypeptide chains:  $\alpha$ ,  $\beta$  and  $\gamma$ . There is evidence for two common alleles at the ADH<sub>2</sub> locus: ADH<sub>2</sub><sup>1</sup> and ADH<sub>2</sub><sup>2</sup>. These alleles code for polypeptides  $\beta^1$  and  $\beta^2$ . There are also two common alleles at the ADH<sub>3</sub> locus: ADH<sub>3</sub><sup>1</sup> and ADH<sub>3</sub><sup>2</sup>, which code for polypeptides  $\gamma^1$  and  $\gamma^2$ . All of the ADH isozymes are dimeric and the subunits interact with each other. In adult liver, all three loci are active. Thus, some of the isozymes are homodimers and some are heterodimers. The heteromeric isozymes contain polypeptides coded by alleles at either the same locus or at different loci. Thus, if an individual has the genotype

ADH<sub>1</sub><sup>1</sup>ADH<sub>1</sub><sup>1</sup>; ADH<sub>2</sub><sup>1</sup>ADH<sub>2</sub><sup>1</sup>; ADH<sub>3</sub><sup>1</sup>ADH<sub>3</sub><sup>2</sup>

the phenotype is  $ADH_1\ 1, ADH_2\ 1, ADH_3\ 2-1$

and in the electrophoretic pattern of a liver extract, there are ten isozymes with the following subunit structures:

|                  |                    |                  |                   |
|------------------|--------------------|------------------|-------------------|
| $\alpha\alpha$   | $\gamma^1\gamma^1$ | $\alpha\gamma^1$ | $\beta^1\gamma^1$ |
| $\alpha\beta^1$  | $\gamma^1\gamma^2$ | $\alpha\gamma^2$ | $\beta^1\gamma^2$ |
| $\beta^1\beta^1$ | $\gamma^2\gamma^2$ |                  |                   |

References

1. Race, R.R. and Sanger, R.: Blood Groups in Man. London: Blackwell, 1975 (6th ed.).
2. Svejgaard, A., Hauge, M., Jersild, C., Platz, P., Ryder, L.P., Nielsen, L.S. and Thomsen, M.: The HLA System: An Introductory Survey. Vol. 7 of Monographs in Human Genetics. Basel: S. Karger, 1976.

In the following table, the enzyme name given is usually that recommended in 1972 by the Enzyme Commission.\* When the E.C. name has not been used as the basis for the symbol, or if another name is much more familiar, the E.C. name is given first, and enclosed in brackets. (In a few instances the E.C. name is not given because it is so similar to the more familiar name.) The locus symbol given first is that recommended by this committee. Alternatives are also listed; these are based on systematic or obsolete names which can nearly always be found in the reference.\* The computer symbols in the table are meant to be initial suggestions; they may require individual revision. The final column indicates that the given locus has been reported to be polymorphic in at least one large ethnic group.

\*Enzyme Nomenclature: Recommendations (1972) of the International Union of Pure and Applied Chemistry and the International Union of Biochemistry.  
Published in 1973 by Elsevier (Amsterdam) and American Elsevier (New York).

Table of Phenotypes for which Information on Chromosomal Assignment is Available

| Name of Phenotype                                | VAM No.†† | E.C. No**<br>(unless<br>not appli-<br>cable = NA) | Locus                  | Alternatives           | Chromosome<br>Assignment<br>or Linkage<br>Group | Computer<br>Symbol‡ | Polymorphic? |
|--|-----------|---|------------------------|------------------------|---|---------------------|--------------|
| ABO blood group                                  | 11030     | NA  | <u>ABO</u>             |                        | 9   | ABO                 | Yes          |
| Acid phosphatase-1                               | 17150     | 3.1.3.2   | <u>AcP1</u>            |                        | 2   | ACP-1               | Yes          |
| Acid phosphatase-2                               | 20095     | 3.1.3.2   | <u>AcP2</u>            |                        | 11  | ACP-2               |              |
| Aconitase, ? mitochondrial                       | 10084     | 4.2.1.3   | <u>Aco</u>             |                        | 3(P)  | ACO                 |              |
| Aconitase, soluble                               | 10085     | 4.2.1.3   | <u>AcoS</u>            |                        | 9(P)  | ACO-S               |              |
| Adenine phosphoribosyltransferase                | 10260     | 2.4.2.7   | <u>APRT</u>            |                        | 16  | APRT                |              |
| Adenosine deaminase                              | 10270     | 3.5.4.4   | <u>ADA</u>             |                        | 20  | ADA                 |              |
| Adenosine kinase                                 | 10275     | 2.7.1.20  | <u>AdK</u>             | <u>AdoK</u>            | 10(P)   | ADK                 |              |
| Adenovirus-12 chromosome<br>modification site-1  | 10293     | NA  | <u>AdV12-CMS-1</u>     |                        | 1   | ADV12-CMS-1         |              |
| Adenovirus-12 chromosome<br>modification site-17 | 10297     | NA  | <u>AdV12-CMS-17</u>    |                        | 17  | ADV12-CMS-17        |              |
| Adenylate kinase-1                               | 10300     | 2.7.4.3   | <u>AK<sub>1</sub></u>  |                        | 9   | AK-1                | Yes          |
| Adenylate kinase-2                               | 10302     | 2.7.4.3   | <u>AK<sub>2</sub></u>  |                        | 1   | AK-2                |              |
| Adenylate kinase-3                               | 10303     | 2.7.4.3   | <u>AK<sub>3</sub></u>  |                        | 9(P)  | AK-3                |              |
| Amylase, pancreatic                              | 10465     | 3.2.1.1   | <u>Amy<sub>2</sub></u> | <u>Amy<sub>P</sub></u> | 1   | AMY-2               | Yes          |
| Amylase, salivary                                | 10470     | 3.2.1.1   | <u>Amy<sub>1</sub></u> | <u>Amy<sub>S</sub></u> | 1   | AMY-1               | Yes          |
| Aniridia, type II(Baltimore)                     | 10620     | NA  |                        |                        | 1(L)  | AN-2                |              |
| $\alpha_1$ -antitrypsin                          | 10740     | ?   | <u>Pi</u>              |                        | 2(I) or<br>12(L)                                | PI                  |              |
| Anti-viral protein                               | 10745     | NA  | <u>AVP</u>             |                        | 21  | AVP                 |              |

\*\* Footnotes on last page.

| Name of Phenotype   | VAM No. | E.C. No.<br>(unless<br>not appli-<br>cable = NA) | Locus                   | Alternatives             | Chromosome<br>Assignment<br>or Linkage<br>Group | Computer<br>Symbol | Polymorphic? |
|---|---------|--|-------------------------|--------------------------|---|--------------------|--------------|
| Auriculo-osteodysplasia<br>B factor (see properdin factor B)                              | 10900   | NA   | <u>AOD</u>              |                          | 1(L)  | AOD                |              |
| Cataract, zonular pulverulent   | 11620   | NA   | <u>Cae</u>              |                          | 1   | CAE                |              |
| Chido blood group   | 11043   | NA   | <u>Ch</u>               |                          | 6   | CH                 |              |
| Citrate synthase, mitochondrial   | 11895   | 4.1.3.7  | <u>CS</u>               |                          | 12(P)   | CS                 |              |
| Complement component-2  | 12060   | NA   | <u>C2</u>               |                          | 6   | C2                 |              |
| Complement component-4  | 12080   | NA   | <u>C4</u>               |                          | 6   | C4                 |              |
| Complement component-8  | 12095   | NA   | <u>C8</u>               |                          | 6   | C8                 |              |
| Desmosterol-to-cholesterol enzyme   | 12565   | ?  | <u>DCE</u>              | <u>D:CE</u>              | 20  | DCE                |              |
| Diphtheria toxin sensitivity  | 12615   | NA   | <u>DTS</u>              |                          | 5(P)  | DTS                |              |
| Dombrock blood group  | 11060   | NA   | <u>Do</u>               |                          | 1(L)  | DO                 |              |
| Duffy blood group   | 11070   | NA   | Fy                      |                          | 1   | FY                 |              |
| Echo 11 sensitivity   | 12915   | NA   | <u>E11S</u>             |                          | 19(L)   | E11S               |              |
| Elliptocytosis-1  | 13050   | NA   | <u>E1</u> <sub>1</sub>  |                          | 1   | E1-1               |              |
| Enolase-1   | 17243   | 4.2.1.11   | <u>Eno</u> <sub>1</sub> | <u>PPH</u> <sub>1</sub>  | 1   | ENO-1              |              |
| Enolase-2   | 13136   | 4.2.1.11   | <u>Eno</u> <sub>2</sub> | <u>PPH</u> <sub>2</sub>  | 12(P)   | ENO-2              |              |
| Esterase activator  | 13325   | ?  |                         |                          | 4(P) or 5                                       | ES-ACT             |              |
| Esterase-A <sub>4</sub>   | 13322   | 3.1.1.1  | <u>EsA</u> <sub>4</sub> | <u>Es-A</u> <sub>4</sub> | 11  | EsA4               |              |
| Esterase D  | 13328   | 3.1.1.1  | <u>EsD</u>              |                          | 13  | ESD                | Yes          |
| Factor B (see properdin factor B)<br>Formylglycinamideribotide<br>(FGAR) amidotransferase | 10255   | ?  | <u>adeB</u>             |                          | 4(P) or 5                                       | ADEB               |              |

| Name of Phenotype                       | VAM No. | E.C. No.<br>(unless<br>not appli-<br>cable = NA) | Locus                          | Alternatives   | Chromosome<br>Assignment<br>or Linkage<br>Group | Computer<br>Symbol | Polymorphic? |
|---|---------|--|--------------------------------|--|---|--------------------|--------------|
| $\alpha$ -L-fucosidase                  | 23000   | 3.2.1.51   | <u><math>\alpha</math> Fuc</u> |  | 1   | A-FUC              | Yes          |
| mitochondrial                           |         |  |                                |  |   |                    |              |
| Fumarate hydratase(fumarase), /         | 13685   | 4.2.1.2  | <u>FH<sub>m</sub></u>          | <u>FH</u>  | 1   | FH-2, FH-M         |              |
| Fumarate hydratase (fumarase),          | 13686   | 4.2.1.2  | <u>FH<sub>s</sub></u>          | <u>FH<sub>1</sub><sup>2</sup></u>                              |   | FH-1, FH-S         |              |
| soluble/                                |         |  |                                |  |   |                    |              |
| Galactokinase                           | 23020   | 2.7.1.6  | <u>Galk</u>                    | <u>GK</u> , <u>GAK</u>   | 17  | GK                 |              |
| Galactose + activator                   | 13703   |  | <u>Gal<sup>+</sup>-Act</u>     |  | 2(P)  |                    |              |
| Galactose-1-phosphate uridylyltrans-    | 23040   | 2.7.7.12   | <u>GalT</u>                    | <u>Gt</u> , <u>Gal-1-PUT</u>                                   | 3   | GPUT, GALT         | Yes          |
| ferase                                  |         |  |                                |  |   |                    |              |
| $\alpha$ -galactosidase (Fabry disease) | 30150   | 3.2.1.22   | <u><math>\alpha</math> Gal</u> |  | X   | A-GAL, A-GAL       |              |
| Glucose-6-phosphate dehydrogenase       | 30590   | 1.1.1.49   | <u>Gd</u>                      | <u>G6PD</u>  | X   | G6PDH, G6PD        | Yes          |
| $\beta$ -Glucuronidase                  | 25322   | 3.2.1.31   | <u><math>\beta</math>Gus</u>   | <u><math>\beta</math>-Glcu</u> , <u><math>\beta</math>-Gcu</u> | 7(I) or 9(I)                                    | GUS, B-GLCU        |              |
| Glutamate- $\gamma$ -semialdehyde       | 13825   | ?  | <u>GSS</u>                     | <u>GSASyt</u> , <u>GSAS</u>                                    | 10(P)   | GSS, GSASYT        |              |
| synthetase                              |         |  |                                |  |   |                    |              |
| Glutamate oxaloacetic transaminase-1    | 13818   | 2.6.1.1  | <u>GOT<sub>S</sub></u>         | <u>GOT-1</u> , <u>GOT<sub>I</sub></u>                          | 10  | GOT-1              |              |
| Glutathione reductase                   | 13830   | 1.6.4.2  | <u>GSR</u>                     |  | 8(P)  | GSR                | Yes          |
| Glyceraldehyde-3-phosphate dehy-        | 13840   | 1.2.1.12   | <u>GAPDH</u>                   | <u>GAPD</u>  | 12(P)   | GAPDH, GAPD        |              |
| drogenase                               |         |  |                                |  |   |                    |              |
| Glyoxylase I                            | 13875   | 4.4.1.5  | <u>GLO<sub>I</sub></u>         | <u>GLY-1</u> , <u>Glx-1</u>                                    | 6   | G-1, GLO-1         | Yes          |
| Gm immunoglobulin types                 | 14710-  | NA   |                                |  | 12 (I)  | Gm                 | Yes          |
| (also see immunoglobulin                | 14719   |  |                                |  |   |                    |              |
| heavy chains)                           |         |  |                                |  |   |                    |              |
| Guanylate kinase-1                      | 13927   | 2.7.4.8  | <u>GuK<sub>1</sub></u>         | <u>Guk<sub>1</sub></u> , <u>GUMPK<sub>1</sub></u>              | 1   | GMPK-1, GUMPK-2    |              |
| Guanylate kinase-2                      | 13928   | 2.7.4.8  | <u>GuK<sub>2</sub></u>         | <u>Guk<sub>2</sub></u> , <u>GUMPK<sub>2</sub></u>              | 1   | GUK-1 & 2          |              |
| Hageman factor                          | 23400   | NA   | <u>HaF</u>                     |  | 7(P)  | HAF                |              |

| Name of Phenotype   | VAM No.                       | E.C. No.<br>(unless<br>not appli-<br>cable = NA) | Locus  | Alternatives                            | Chromosome<br>Assignment<br>or Linkage<br>Group | Computer<br>Symbol | Polymorphic? |
|---|-------------------------------|--|--|---|---|--------------------|--------------|
| Haptoglobin, alpha  | 14010                         | NA   | <u>Hp</u>  |   | 16  | A-HP               |              |
| Hemoglobin, alpha or beta   | 14180, 14190                  | NA   | <u>Hb</u> , <u>Hb</u>                                      |   | 2(I) & 4(I)                                     | A-HB, B-HB         |              |
| Hexokinase-1  | 14260                         | 2.7.1.1  | <u>Hk</u> <sub>1</sub>                                     | <u>Hk</u> <sub>I</sub>                  | 10  | HK-1               |              |
| Hexosaminidase A  | 27280                         | 3.2.1.30   | <u>Hex</u> <sub>A</sub>                                    | <u>NAGA</u> <sub>A</sub>                | 15  | Hex-A, Hex A       |              |
| Hexosaminidase B  | 14265                         | 3.2.1.30   | <u>Hex</u> <sub>B</sub>                                    | <u>NAGA</u> <sub>B</sub>                | 5   | Hex-B, Hex B       |              |
| HLA: Major histocompatibility complex                             | 14280, 14283,<br>14284, 15785 | NA   | <u>HLA-A</u> , <u>HLA-B</u><br><u>HLA-C</u> , <u>HLA-D</u> | (Several)                               | 6   | HLA                |              |
| Hypoxanthine-guanine phosphoribosyltransferase                    | 30800                         | 2.4.2.8  | <u>HPRT</u>  |   | X   | HGPRT              |              |
| Immune response   | 14685                         | NA   | <u>Ir</u>  |   | 6(L)  | IR                 |              |
| Immunoglobulin heavy chains<br>(also see Gm immunoglobulin types) | 14710-14719                   | NA   |  |   | 2(I)  | Ig                 |              |
| Indophenoloxidase (see superoxide dismutase)                      |                               |  |  |   |   |                    |              |
| Inosine triphosphatase  | 14753                         | 3.6.1.19   | <u>ITP</u>   |   | 20  | ITP                |              |
| Interferon-1  | 14757                         | NA   | <u>If</u> <sub>1</sub>                                     |   | 2(P)  | IF-1               |              |
| Interferon-2  | 14758                         | NA   | <u>If</u> <sub>2</sub>                                     |   | 5(P)  | IF-2               |              |
| Isocitrate dehydrogenase-1  | 14770                         | 1.1.1.42   | <u>ICDH</u> <sub>S</sub>                                   | <u>IDH</u> <sub>S</sub> , <u>IDH</u> -1 | 2   | ICDH-1, IDH-1      |              |
| Isocitrate dehydrogenase, mitochondrial                           | 14765                         | 1.1.1.42   | <u>ICDH</u> <sub>M</sub>                                   | <u>IDH</u> <sub>M</sub> , <u>IDH</u> -2 | 15(P)   | ICDH-2, IDH-M      |              |
| Lactate dehydrogenase A   | 15000                         | 1.1.1.27   | <u>LDH</u> <sub>A</sub>                                    | <u>LDH</u> -A                           | 11  | LDH-A              |              |
| Lactate dehydrogenase B   | 15010                         | 1.1.1.27   | <u>LDH</u> <sub>B</sub>                                    | <u>LDH</u> -B                           | 12  | LDH-B              |              |
| Lecithin-cholesterol acyltransferase                              | 24590                         | 2.3.1.43   | <u>LCAT</u>  |   | 15  | LCAT               |              |

| Name of Phenotype                   | VAM No.     | E.C. No.<br>(unless<br>not appli-<br>cable = NA) | Locus   | Alternatives  | Chromosome<br>Assignment<br>or Linkage<br>Group | Computer<br>Symbol | Polymorphic? |
|-------------------------------------|-------------|--|---|---|---|--------------------|--------------|
| Lethal antigen                      | 15125-15127 | NA   | <u>a</u> <sub>1</sub> , <u>a</u> <sub>2</sub> , <u>a</u> <sub>3</sub> |   | 11  | AL                 |              |
| Malate dehydrogenase-1              | 15420       | 1.1.1.37   | <u>MDH</u> <sub>S</sub>   | <u>MOR</u> <sub>S</sub> , <u>MOR</u> -1,<br><u>MDH</u> -1 | 2   | MDH-1              |              |
| Malate dehydrogenase, mitochondrial | 15410       | 1.1.1.37   | <u>MDH</u> <sub>M</sub>   | <u>MOR</u> <sub>M</sub> , <u>MOR</u> -2,<br><u>MDH</u> -2 | 7   | MDH-2              |              |
| Malic enzyme-1                      | 15425       | 1.1.1.40   | <u>ME</u> <sub>S</sub>  | <u>MOD</u> <sub>S</sub> , <u>MOD</u> -1,<br><u>ME</u> -1  | 6   | ME-1               |              |
| Mannosephosphate isomerase          | 15455       | 5.3.1.8  | <u>MPI</u>  |   | 15  | MANPI, MPI         |              |
| $\beta$ 2-microglobulin             | 10970       | NA   | <u><math>\beta</math>2M</u>   |   | 15  | B-2M               |              |
| MNSs blood group                    | 11130       | NA   | <u>MNSs</u>   |   | 2(L)  | MNS                |              |
| Nail-patella syndrome               | 16120       | NA   | <u>NPA</u>  | <u>NP</u>   | 9   | NPA                |              |
| Nucleoside phosphorylase            | 16405       | 2.4.2.1  | <u>NP</u>   |   | 14  | NP                 |              |
| P blood group                       | 11140       | NA   | <u>P</u>  |   | 6(L)  | P                  |              |
| Pepsinogen                          | 16970       | 3.4.23*  | <u>Pg</u>   | <u>Pg</u> -5  | 6   | PEPSG, Puc         | Yes          |
| Peptidase A                         | 16980       | 3.4.11.*   | <u>Pep</u> A  |   | 18  | PEPA               | Yes          |
| Peptidase B                         | 16990       | 3.4.11.*   | <u>Pep</u> B  |   | 12  | PEPB               |              |
| Peptidase C                         | 17000       | 3.4.11.*   | <u>Pep</u> C  |   | 1   | PEPC               | Yes          |
| Peptidase D                         | 17010       | 3.4.13.9   | <u>Pep</u> D  |   | 19(P)   | PEPD               | Yes          |
| Phosphoglucomutase-1                | 17190       | 2.7.5.1  | <u>PGM</u> <sub>1</sub>   |   | 1   | PGM-1              | Yes          |
| Phosphoglucomutase-2                | 17200       | 2.7.5.1  | <u>PGM</u> <sub>2</sub>   |   | 4(P)  | PGM-2              | Yes          |
| Phosphoglucomutase-3                | 17210       | 2.7.5.1  | <u>PGM</u> <sub>3</sub>   |   | 6   | PGM-3              | Yes          |

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|--|---------|--|------------------------|---|---|--------------------|--------------|
| 6-phosphogluconate dehydrogenase   | 17220   | 1.1.1.44   | <u>PGD</u>             | <u>6PGD</u>                                     | 1   | 6PGD               | Yes          |
| Phosphoglycerate kinase  | 31180   | 2.7.2.3  | <u>PGK</u>             |   | X   | PGAK, PGK          |              |
| Phosphohexose isomerase  | 17240   |  | <u>PHI</u>             |   | 19  | PHI                |              |
| Phosphopyruvate hydratase (see enolase)                                    |         |  |                        |   |   |                    |              |
| Phosphoribosyl glycineamide<br>synthetase                                  | 13844   | 6.3.4.13   | <u>GAPS</u>            |   | 21(P)   | GAPS               |              |
| Polio sensitivity  | 17385   | NA   | <u>PVS</u>             |   | 19  | PVS                |              |
| Properdin factor B   | 13847   | NA   | <u>Bf</u>              |   | 6   | B                  |              |
| Pyrophosphatase, inorganic   | 17903   | 3.6.1.1  | <u>PP</u>              |   | 10  | PP                 |              |
| Pyruvate kinase-3 (M2)   | 17905   | 2.7.1.40   | <u>PK<sub>M2</sub></u> | <u>PK<sub>III</sub></u> , <u>PK<sub>3</sub></u> | 15  | PK-M2, PK3         |              |
| Retinoblastoma-1   | 18020   | NA   | <u>Rb<sub>1</sub></u>  |   | 13(L)   | RB <sub>1</sub>    |              |
| Rhesus blood group   | 11170   | NA   | <u>Rh</u>              |   | 1   | RH                 |              |
| Ribosomal RNA  | 18045   | NA   | NA                     |   | 13, 14, 15,<br>21, 22                           | R-RNA              |              |
| Rodgers blood group  | 11171   | NA   | <u>Rg</u>              |   | 6   | RG                 |              |
| 5S RNA gene(s)   | 18042   | NA   | <u>RN5S</u>            |   | 1   | RN5S               |              |
| Scianna blood group  | 11175   | NA   | <u>Sc</u>              |   | 1(L)  | SC                 |              |
| Sclerotylosis  | 18160   | NA   | <u>Tys</u>             |   | 2(L)  | TYS                |              |
| Serine hydroxymethyltransferase<br>(glycine + A auxotroph comple-<br>ting) | 13845   | 2.1.2.1  | <u>SHMT</u>            |   | 12(P)   | SHMT               |              |
| Spherocytosis, Denver type   | 18290   | NA   | <u>Sph<sub>1</sub></u> |   | 8(L) or 12(L)                                   | Sph-1              |              |

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|---|---------|--|-------------------------|--|---|---------------------|--------------|
| Superoxide dismutase-1                            | 14745   | 1.15.1.1   | <u>SOD</u> <sub>S</sub> | <u>IPO</u> -A, <u>SOD</u> -A,<br><u>SOD</u> -1 | 21  | SOD-1               |              |
| Superoxide dismutase-2                            | 14746   | 1.15.1.1   | <u>SOD</u> <sub>M</sub> | <u>IPO</u> -B, <u>SOD</u> -B,<br><u>SOD</u> -2 | 6   | SOD-2               |              |
| SV <sup>40</sup> -T antigen                       | 18680   | NA   | <u>SV40-T</u>           |  | 7(P)  | SV <sup>40</sup> -T |              |
| Testis determining factor                         | --      | NA   | <u>TDF</u>              |  | Y   | TDF                 |              |
| Thymidine kinase, mitochondrial                   | 18829   | 2.7.1.75   | <u>TK</u> <sub>m</sub>  | <u>TK</u> <sub>2</sub>                         | 16(P)   | TK <sub>m</sub>     |              |
| Thymidine kinase, soluble                         | 18830   | 2.7.1.75   | <u>TK</u> <sub>s</sub>  | <u>TK</u> <sub>1</sub>                         | 17  | TK <sub>s</sub>     |              |
| Trisephosphate isomerase                          | 19045   | 5.3.1.1  | <u>TPI</u>              |  | 12  | TPI                 |              |
| Tryptophanyl-tRNA synthetase                      | 19105   | 6.1.1.2  | <u>TrpRS</u>            |  | 14  | TRPRS               |              |
| Uridyl diphosphate glucose pyro-<br>phosphorylase | 19175   | 2.7.7.9  | <u>UGPP</u>             |  | 1(P)  | UGPP                |              |
| Waardenburg syndrome                              | 19350   | NA   | <u>WS</u> <sub>1</sub>  |  | 9(L)  | WS-1                |              |
| Xeroderma pigmentosum, Egyptian                   | 27870   | NA   | <u>XP</u> <sub>E</sub>  |  | 9(L)  | XP-E                |              |
| X-linked species (or surface)<br>antigen          | 31345   | NA   | <u>SAX</u>              |  | X   | SAX                 |              |
| Y histocompatibility antigen                      | --      | NA   | <u>H-Y</u>              |  | Y   | H-Y                 |              |

\* Enzyme Nomenclature: Recommendations (1972) of the International Union of Pure and Applied Chemistry and the International Union of Biochemistry, 1973, Elsevier (Amsterdam) and American Elsevier (New York).

† When within the capability of the computer, lower case should be used as in the locus symbols.

†† Number assigned to locus in McKusick's Mendelian Inheritance in Man (4th ed., 1975 with additions).